Medical curriculum vitae of a man who was confronted relatively late with his Primary Hyperoxaluria Type 1 disease

- 40 years of completely normal life

- Between 40 - 50 years of age there were 2 renal colics. In both cases, it was neglected to have the kidney stones examined for their composition. This was the first "cardinals" mistake, because an examination would have very probably given clear indications (too high oxalate values) of my PH1 disease.

- When I was 57, I had a colon cancer operation, in which about 35 cm of the rectum was removed. In addition, an artificial outlet was created (was put back after 6 weeks). Concerning the cancer everything went well and positive, but shortly after the intestinal surgery my kidney values already deteriorated during the inpatient follow-up treatment. The doctors saw the deterioration, but initially classified it as a normal side effect of the operation. Although I drank 2-3 liters of water daily, my kidney values continued to deteriorate, certainly also because I lost a lot of fluid through the artificial outlet, which the kidneys then lacked for "flushing". The doctors considered a kidney biopsy, but it was not followed up. This was the second "Cardinals" mistake, because at that time one could have got the information about too high oxalate values in time. After a 3-month hospital stay, the doctors did not find reason for my high kidney values (approx. 7 mg/dl creatinine) and I was referred to a nephrologist in an emergency situation.

- The nephrologist performed a kidney biopsy after one week of preliminary examinations. After another week, the evaluation showed huge amounts of oxalate deposits and told the disease "primary hyperoxaluria". My creatinine values had worsened in the meantime (far more than 10 mg/dl), so that I became a "dialysis" patient from then on.

- With the very rare disease "Primary Hyperoxaluria" (1 in 1 million) the nephrologists were also overtaxed and I was referred to the Children's hospital of the University of Bonn, because there is/was a department especially for this disease (recently moved to the Children's Kidney Centre in Bonn). The extensive examinations (incl. genetic analysis) showed that my kidney failure is due to a congenital metabolic disease which produces too much oxalate in the liver and is called "primary hyperoxaluria". There are 3 different types and I have type 1 (most common type). This congenital gene defect could also be present in my 3 siblings and their examinations showed that one of my brothers also had PH Type1 and could therefore be saved from kidney failure for the time being. About 25-50% of the siblings are usually affected. Although there is hardly any medication available to treat this disease, I am very lucky to limit the oxalate deposits in my body by taking high amounts of vitamin B6 in such a way that I hardly have any further deposits (by the way, it also helps with my brother). This is the only reason why I am now registered as a "transplant label" with Eurotransplant and can hope that I can now expect a transplant of a new kidney in the 7th year of dialysis.

Recommendation: Pay attention to the analysis of kidney stones, so that a PH treatment can take place in time.